



## tetrasomy 18p

Tetrasomy 18p is a chromosomal condition that affects many parts of the body. This condition usually causes feeding difficulties in infancy, delayed development, intellectual disability that is often mild to moderate but can be severe, changes in muscle tone, distinctive facial features, and other birth defects. However, the signs and symptoms vary among affected individuals.

Babies with tetrasomy 18p often have trouble feeding and may vomit frequently, which makes it difficult for them to gain weight. Some affected infants also have breathing problems and jaundice, which is a yellowing of the skin and the whites of the eyes.

Changes in muscle tone are commonly seen with tetrasomy 18p. Some affected children have weak muscle tone (hypotonia), while others have increased muscle tone (hypertonia) and stiffness (spasticity). These changes contribute to delayed development of motor skills, including sitting, crawling, and walking.

Tetrasomy 18p is associated with a distinctive facial appearance that can include unusually shaped and low-set ears, a small mouth, a flat area between the upper lip and the nose (philtrum), and a thin upper lip. Many affected individuals also have a high, arched roof of the mouth (palate), and a few have had a split in the roof of the mouth (cleft palate).

Additional features of tetrasomy 18p can include seizures, vision problems, recurrent ear infections, mild to moderate hearing loss, constipation and other gastrointestinal problems, abnormal curvature of the spine (scoliosis or kyphosis), a shortage of growth hormone, and birth defects affecting the heart and other organs. Males with tetrasomy 18p may be born with undescended testes (cryptorchidism) or the opening of the urethra on the underside of the penis (hypospadias). Psychiatric conditions, such as attention deficit hyperactivity disorder (ADHD) and anxiety, as well as social and behavioral challenges have also been reported in some people with tetrasomy 18p.

### Frequency

Tetrasomy 18p is a rare disorder. It is known to affect about 250 families worldwide.

### Genetic Changes

Tetrasomy 18p results from the presence of an abnormal extra chromosome, called an isochromosome 18p, in each cell. An isochromosome is a chromosome with two identical arms. Normal chromosomes have one long (q) arm and one short (p) arm, but isochromosomes have either two q arms or two p arms. Isochromosome 18p is a version of chromosome 18 made up of two p arms.

Cells normally have two copies of each chromosome, one inherited from each parent. In people with tetrasomy 18p, cells have the usual two copies of chromosome 18 plus an isochromosome 18p. As a result, each cell has four copies of the short arm of chromosome 18. (The word "tetrasomy" is derived from "tetra," the Greek word for "four.") The extra genetic material from the isochromosome disrupts the normal course of development, causing the characteristic features of this disorder.

## **Inheritance Pattern**

Tetrasomy 18p is usually not inherited. The chromosomal change responsible for the disorder typically occurs as a random event during the formation of reproductive cells (eggs or sperm) in a parent of the affected individual, usually the mother. Most affected individuals have no history of the disorder in their family. However, rare inherited cases of tetrasomy 18p have been reported.

## **Other Names for This Condition**

- 18p isochromosome
- 18p tetrasomy

## **Diagnosis & Management**

### Genetic Testing

- Genetic Testing Registry: Chromosome 18, tetrasomy 18p  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0795868/>

### Other Diagnosis and Management Resources

- Chromosome 18 Clinical Research Center, University of Texas Health Science Center at San Antonio  
<http://www.pediatrics.uthscsa.edu/centers/chromosome18/>

### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

## **Additional Information & Resources**

### MedlinePlus

- Encyclopedia: Chromosome  
<https://medlineplus.gov/ency/article/002327.htm>
- Health Topic: Developmental Disabilities  
<https://medlineplus.gov/developmentaldisabilities.html>

### Genetic and Rare Diseases Information Center

- Chromosome 18p tetrasomy  
<https://rarediseases.info.nih.gov/diseases/35/chromosome-18p-tetrasomy>

### Educational Resources

- Disease InfoSearch: Chromosome 18, Tetrasomy 18p  
<http://www.diseaseinfosearch.org/Chromosome+18%2C+Tetrasomy+18p/1484>
- MalaCards: chromosome 18p tetrasomy  
[http://www.malacards.org/card/chromosome\\_18p\\_tetrasomy](http://www.malacards.org/card/chromosome_18p_tetrasomy)
- March of Dimes: Chromosomal Conditions  
<http://www.marchofdimes.org/baby/chromosomal-conditions.aspx>
- Orphanet: Tetrasomy 18p  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=3307](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=3307)

### Patient Support and Advocacy Resources

- Chromosome 18 Registry & Research Society  
<https://www.chromosome18.org/tetrasomy-18p/>
- Chromosome Disorder Outreach  
<http://chromodisorder.org/>
- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/chromosome-18-tetrasomy-18p/>
- Resource list from the University of Kansas Medical Center: Chromosomal Conditions  
<http://www.kumc.edu/gec/support/chromoso.html>
- Unique - The Rare Chromosome Disorder Support Group (UK)  
<http://www.rarechromo.org/>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28tetrasomy+18p%5BTIAB%5D%29+OR+%28isochromosome+18p%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## **Sources for This Summary**

- Bugge M, Blennow E, Friedrich U, Petersen MB, Pedetour F, Tsezou A, Orum A, Hermann S, Lyngbye T, Sarri C, Avramopoulos D, Kitsiou S, Lambert JC, Guzda M, Tommerup N, Brøndum-Nielsen K. Tetrasomy 18p de novo: parental origin and different mechanisms of formation. *Eur J Hum Genet.* 1996;4(3):160-7. Erratum in: *Eur J Hum Genet* 1996;4(5):291.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/8840116>
- Nucaro A, Chillotti I, Pisano T, Pruna D, Cianchetti C. Progressive spastic paraplegia as a feature of tetrasomy 18p. *Am J Med Genet A.* 2010 Sep;152A(9):2173-5. doi: 10.1002/ajmg.a.33576.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20684001>
- O'Donnell L, Soileau BT, Sebold C, Gelfond J, Hale DE, Cody JD. Tetrasomy 18p: report of cognitive and behavioral characteristics. *Am J Med Genet A.* 2015 Jul;167(7):1474-82. doi: 10.1002/ajmg.a.37036. Epub 2015 Apr 21.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25900901>
- Sebold C, Roeder E, Zimmerman M, Soileau B, Heard P, Carter E, Schatz M, White WA, Perry B, Reinker K, O'Donnell L, Lancaster J, Li J, Hasi M, Hill A, Pankratz L, Hale DE, Cody JD. Tetrasomy 18p: report of the molecular and clinical findings of 43 individuals. *Am J Med Genet A.* 2010 Sep; 152A(9):2164-72. doi: 10.1002/ajmg.a.33597.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20803640>
- Soileau B, Hasi M, Sebold C, Hill A, O'Donnell L, Hale DE, Cody JD. Adults with Chromosome 18 Abnormalities. *J Genet Couns.* 2015 Aug;24(4):663-74. doi: 10.1007/s10897-014-9793-5. Epub 2014 Nov 19.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25403900>
- Zavala J, Ramirez M, Medina R, Heard P, Carter E, Crandall A, Hale D, Cody J, Escamilla M. Psychiatric syndromes in individuals with chromosome 18 abnormalities. *Am J Med Genet B Neuropsychiatr Genet.* 2010 Apr 5;153B(3):837-45. doi: 10.1002/ajmg.b.31047.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19927307>

---

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/tetrasomy-18p>

Reviewed: April 2016

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services